

**Table 3. Summary of Sequence Variations in ZIC2**

Mutations	Sequence Change	Amino Acid Location	Type of Mutation	Reference
	1 bp del G	7	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	56 bp ins	60	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	1 bp del G	312	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	7 bp del	348	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	2 bp del AG	364	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	1 bp ins C	440	Frameshift	<a href="#">Brown et al 1998, 2001</a>
	30 bp ins	468-478	Alanine expansion <sup>1</sup>	<a href="#">Brown et al 1998, 2001</a>

1. Alanine expansion found in five unrelated individuals

## References

Brown SA, Odent S, David V, Blayau M, Dubourg C, Apacik C, Delgado MA, Hall BD, Reynolds JF, Sommer A, Wieczorek D, Brown SA, Muenke M (2001) Holoprosencephaly due to mutations in ZIC2: alanine tract expansion mutations may be caused by parental somatic recombination. *Hum Mol Genet* 10:791-6 [[Medline](#)]

Brown SA, Warburton D, Brown LY, Yu CY, Roeder ER, Stengel-Rutkowski S, Hennekam RC, Muenke M (1998) Holoprosencephaly due to mutations in ZIC2, a homologue of Drosophila odd-paired. *Nat Genet* 20:180-3 [[Medline](#)]